Nadia Chuzhanova

List of Publications by Year
in descending order

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65 papers	5,736 citations	136740 32 h-index	106150 65 g-index
68 all docs	68 docs citations	68 times ranked	8288 citing authors

#	Article	IF	Citations
1	Predicting novel genomic regions linked to genetic disorders using GWAS and chromosome conformation data â€“ a case study of schizophrenia. Scientific Reports, 2019, 9, 17940.	1.6	6
2	Identification of novel genes associated with longevity in Drosophila melanogaster - a computational approach. Aging, 2019, 11, 11244-11267.	1.4	6
3	Complexity and robustness in hypernetwork models of metabolism. Journal of Theoretical Biology, 2016, 406, 99-104.	0.8	16
4	A Role for Non-B DNA Forming Sequences in Mediating Microlesions Causing Human Inherited Disease. Human Mutation, 2016, 37, 65-73.	1.1	22
5	Network motif frequency vectors reveal evolving metabolic network organisation. Molecular BioSystems, 2015, 11, 77-85.	2.9	7
6	Remotely acting SMCHD1 gene regulatory elements: in silico prediction and identification of potential regulatory variants in patients with FSHD. Human Genomics, 2015, 9, 25.	1.4	0
7	Screening in silico predicted remotely acting NF1gene regulatory elements for mutations in patients with neurofibromatosis type 1. Human Genomics, 2013, 7, 18.	1.4	4
8	Ornithine carbamoyltransferase deficiency: molecular characterization of 29 families. Clinical Genetics, 2013, 84, 552-559.	1.0	6
9	Exploring the somatic NF1 mutational spectrum associated with NF1 cutaneous neurofibromas. European Journal of Human Genetics, 2012, 20, 411-419.	1.4	25
10	Comparative Analysis of Genome Sequences Covering the Seven Cronobacter Species. PLoS ONE, 2012, 7, e49455.	1.1	130
11	Identification of recurrent type-2NF1microdeletions reveals a mitotic nonallelic homologous recombination hotspot underlying a human genomic disorder. Human Mutation, 2012, 33, 1599-1609.	1.1	26
12	Characterization of the nonallelic homologous recombination hotspot PRS3 associated with type-3NF1deletions. Human Mutation, 2012, 33, 372-383.	1.1	28
13	The Somatic Mutational Spectrum of the NF1 Gene. , 2012, , 211-233.		3
14	In Silico identification of pathogenic strains of Cronobacter from Biochemical data reveals association of inositol fermentation with pathogenicity. BMC Microbiology, 2011, 11, 204.	1.3	20
15	Comparative analysis of germline and somatic microlesion mutational spectra in 17 human tumor suppressor genes. Human Mutation, 2011, 32, 620-632.	1.1	13
16	Non-B DNA-forming Sequences and WRN Deficiency Independently Increase the Frequency of Base Substitution in Human Cells. Journal of Biological Chemistry, 2011, 286, 10017-10026.	1.6	31
17	An isolated case of lissencephaly caused by the insertion of a mitochondrial genome-derived DNA sequence into the 5' untranslated region of the PAFAH1B1 (LIS1) gene. Human Genomics, 2010, 4, 384.	1.4	10
18	Analysis of NF1 somatic mutations in cutaneous neurofibromas from patients with high tumor burden. Neurogenetics, 2010, 11, 391-400.	0.7	25

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19	Complete ascertainment of intragenic copy number mutations (CNMs) in the CFTR gene and its implications for CNM formation at other autosomal loci. Human Mutation, 2010, 31, 421-428.	1.1	31
20	A novel third type of recurrent NF1 microdeletion mediated by nonallelic homologous recombination between LRRC37B-containing low-copy repeats in 17q11.2. Human Mutation, 2010, 31, 742-751.	1.1	42
21	Genes, mutations, and human inherited disease at the dawn of the age of personalized genomics. Human Mutation, 2010, 31, 631-655.	1.1	161
22	Intrachromosomal mitotic nonallelic homologous recombination is the major molecular mechanism underlying type-2 NF1 deletions. Human Mutation, 2010, 31, 1163-1173.	1.1	36
23	SPRED1 mutations (Legius syndrome): another clinically useful genotype for dissecting the neurofibromatosis type 1 phenotype. Journal of Medical Genetics, 2009, 46, 431-437.	1.5	83
24	A gene conversion hotspot in the human growth hormone (GH1) gene promoter. Human Mutation, 2009, 30, 239-247.	1.1	13
25	Identification and molecular characterization of six novel mutations in the UDP-N-acetylglucosamine-1-phosphotransferase gamma subunit (GNPTG) gene in patients with mucolipidosis III gamma. Human Mutation, 2009, 30, 978-984.	1.1	26
26	Gene conversion causing human inherited disease: Evidence for involvement of non-B-DNA-forming sequences and recombination-promoting motifs in DNA breakage and repair. Human Mutation, 2009, 30, 1189-1198.	1.1	63
27	Molecular characterization of 22 novel UDP-N-acetylglucosamine-1-phosphate transferase Î±- and Î²-subunit (GNPTAB) gene mutations causing mucolipidosis types IIÎ±/Î² and IIIÎ±/Î² in 46 patients. Human Mutation, 2009, 30, E956-E973.	1.1	38
28	Genomeâ€wide highâ€resolution analysis of DNA copy number alterations in NF1â€associated malignant peripheral nerve sheath tumors using 32K BAC array. Genes Chromosomes and Cancer, 2009, 48, 897-907.	1.5	50
29	The spectrum of somatic and germline NF1 mutations in NF1 patients with spinal neurofibromas. Neurogenetics, 2009, 10, 251-263.	0.7	61
30	Cruciform-forming inverted repeats appear to have mediated many of the microinversions that distinguish the human and chimpanzee genomes. Chromosome Research, 2009, 17, 469-483.	1.0	31
31	Germline and somaticNF1 gene mutation spectrum in NF1-associated malignant peripheral nerve sheath tumors (MPNSTs). Human Mutation, 2008, 29, 74-82.	1.1	106
32	A meta-analysis of nonsense mutations causing human genetic disease. Human Mutation, 2008, 29, 1037-1047.	1.1	348
33	Two sisters with Rett syndrome and non-identical paternally-derived microdeletions in the MECP2 gene. Genomic Medicine, 2008, 2, 77-81.	0.6	4
34	High-Resolution DNA Copy Number Profiling of Malignant Peripheral Nerve Sheath Tumors Using Targeted Microarray-Based Comparative Genomic Hybridization. Clinical Cancer Research, 2008, 14, 1015-1024.	3.2	119
35	Co-inheritance of a novel deletion of the entire SPINK1 gene with a CFTR missense mutation (L997F) in a family with chronic pancreatitis. Molecular Genetics and Metabolism, 2007, 92, 168-175.	0.5	25
36	Gain-of-glycosylation mutations. Current Opinion in Genetics and Development, 2007, 17, 245-251.	1.5	65

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37	An Absence of Cutaneous Neurofibromas Associated with a 3-bp Inframe Deletion in Exon 17 of the NF1 Gene (c.2970-2972 delAAT): Evidence of a Clinically Significant NF1 Genotype-Phenotype Correlation. American Journal of Human Genetics, 2007, 80, 140-151.	2.6	335
38	Type 2 NF1 Deletions Are Highly Unusual by Virtue of the Absence of Nonallelic Homologous Recombination Hotspots and an Apparent Preference for Female Mitotic Recombination. American Journal of Human Genetics, 2007, 81, 1201-1220.	2.6	60
39	Gene conversion: mechanisms, evolution and human disease. Nature Reviews Genetics, 2007, 8, 762-775.	7.7	576
40	Searching for potential microRNA-binding site mutations amongst known disease-associated 3â€² UTR variants. Genomic Medicine, 2007, 1, 29-33.	0.6	7
41	A novel Alu-mediated 61-kb deletion of the von Willebrand factor (VWF) gene whose breakpoints co-locate with putative matrix attachment regions. Blood Cells, Molecules, and Diseases, 2006, 36, 385-391.	0.6	19
42	A novel gross deletion caused by non-homologous recombination of the PDHX gene in a patient with pyruvate dehydrogenase deficiencyâ˜†. Molecular Genetics and Metabolism, 2006, 89, 106-110.	0.5	13
43	Gross genomic rearrangements involving deletions in the CFTR gene: characterization of six new events from a large cohort of hitherto unidentified cystic fibrosis chromosomes and meta-analysis of the underlying mechanisms. European Journal of Human Genetics, 2006, 14, 567-576.	1.4	77
44	Long homopurine*homopyrimidine sequences are characteristic of genes expressed in brain and the pseudoautosomal region. Nucleic Acids Research, 2006, 34, 2663-2675.	6.5	60
45	New Screening Software Shows that Most Recent Large 16S rRNA Gene Clone Libraries Contain Chimeras. Applied and Environmental Microbiology, 2006, 72, 5734-5741.	1.4	621
46	Metaâ€Analysis of gross insertions causing human genetic disease: Novel mutational mechanisms and the role of replication slippage. Human Mutation, 2005, 25, 207-221.	1.1	148
47	Gains of glycosylation comprise an unexpectedly large group of pathogenic mutations. Nature Genetics, 2005, 37, 692-700.	9.4	198
48	Breakpoint analysis of the pericentric inversion distinguishing human chromosome 4 from the homologous chromosome in the chimpanzee (Pan troglodytes). Human Mutation, 2005, 25, 45-55.	1.1	47
49	Complex gene rearrangements caused by serial replication slippage. Human Mutation, 2005, 26, 125-134.	1.1	88
50	Microdeletions and microinsertions causing human genetic disease: common mechanisms of mutagenesis and the role of local DNA sequence complexity. Human Mutation, 2005, 26, 205-213.	1.1	136
51	Intrachromosomal serial replication slippage intransgives rise to diverse genomic rearrangements involving inversions. Human Mutation, 2005, 26, 362-373.	1.1	62
52	Molecular characterisation of the pericentric inversion that distinguishes human chromosome 5 from the homologous chimpanzee chromosome. Human Genetics, 2005, 117, 168-176.	1.8	27
53	At Least 1 in 20 16S rRNA Sequence Records Currently Held in Public Repositories Is Estimated To Contain Substantial Anomalies. Applied and Environmental Microbiology, 2005, 71, 7724-7736.	1.4	716
54	Independent intrachromosomal recombination events underlie the pericentric inversions of chimpanzee and gorilla chromosomes homologous to human chromosome 16. Genome Research, 2005, 15, 1232-1242.	2.4	42

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55	Breakpoints of gross deletions coincide with non-B DNA conformations. Proceedings of the National Academy of Sciences of the United States of America, 2004, 101, 14162-14167.	3.3	184
56	Genomic rearrangements in theCFTRgene: Extensive allelic heterogeneity and diverse mutational mechanisms. Human Mutation, 2004, 23, 343-357.	1.1	115
57	Indel in the FIC1/ATP8B1 gene?a novel rare type of mutation associated with benign recurrent intrahepatic cholestasis. Hepatology Research, 2004, 30, 1-3.	1.8	1
58	Meta-analysis of indels causing human genetic disease: mechanisms of mutagenesis and the role of local DNA sequence complexity. Human Mutation, 2003, 21, 28-44.	1.1	112
59	Translocation and gross deletion breakpoints in human inherited disease and cancer II: Potential involvement of repetitive sequence elements in secondary structure formation between DNA ends. Human Mutation, 2003, 22, 245-251.	1.1	98
60	Translocation and gross deletion breakpoints in human inherited disease and cancer I: Nucleotide composition and recombination-associated motifs. Human Mutation, 2003, 22, 229-244.	1.1	214
61	A rare complex DNA rearrangement in the murine Steel gene results in exon duplication and a lethal phenotype. Blood, 2003, 102, 3548-3555.	0.6	5
62	Identification of an intronic regulatory element in the human protein C (PROC) gene. Human Genetics, 2000, 107, 458-465.	1.8	20
63	Changes in primary DNA sequence complexity influence the phenotypic consequences of mutations in human gene regulatory regions. Human Genetics, 2000, 107, 362-365.	1.8	16
64	Promoter shuffling has occurred during the evolution of the vertebrate growth hormone gene. Gene, 2000, 254, 9-18.	1.0	19
65	Evolution of the proximal promoter region of the mammalian growth hormone gene. Gene, 1999, 237, 143-151.	1.0	34